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ABSTRACT

The document describes Lowe's syndrome, a hereditary condition that affects only males and is typically diagnosed during the first year of life. Effects of Lowe's syndrome on the eyes (cataracts, glaucoma, corneal degeneration, and strabismus) are discussed, as well as related problems with the central nervous system, muscles, kidneys, bones, and teeth. Special health concerns arising from these effects include metabolic imbalance, respiratory illness, and constipation. Additional topics addressed in the guide are prognosis, related information on genetics (including females as carriers and genetic counseling), ongoing research efforts, developmental and educational programming, and family dynamics. The booklet concludes with a brief description of Lowe's Syndrome Association, a nonprofit organization of parents and professionals founded in 1983. (JW)

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Living With Lowe's Syndrome

A Guide for Families, Friends, and Professionals

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Living With Lowe's Syndrome

A Guide for Families, Friends, and Professionals

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Lowe's Syndrome Association, Inc.
West Lafayette, Indiana
1987

Foreword

Learning that their child has Lowe's syndrome is a shocking and devastating experience for most parents. Most feel unprepared and ill-equipped for the challenges of raising a child with such complex medical and educational needs. Some eventually discover many resources available to help them, including the support of family and friends, community programs, and excellent medical care.

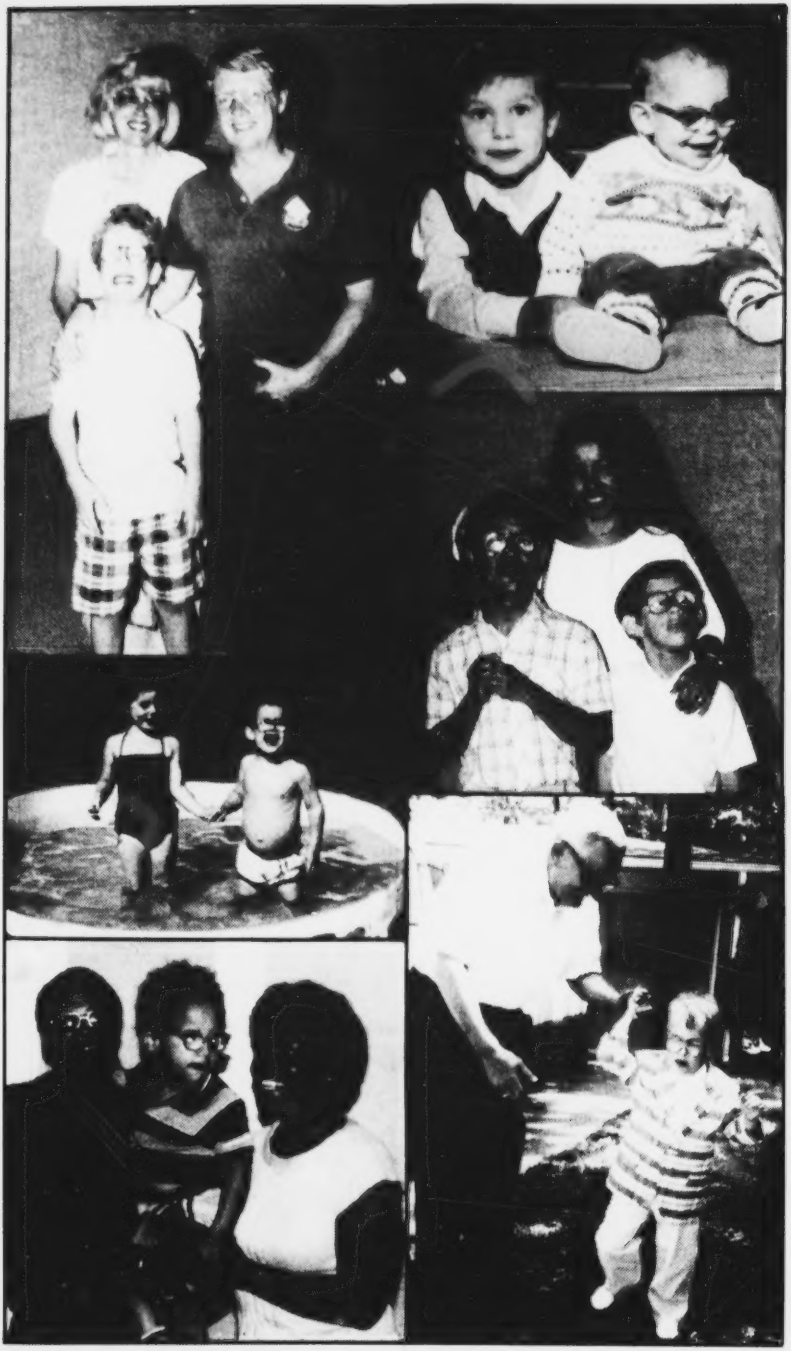
Most, however, will be frustrated in their efforts to obtain accurate and up-to-date information on their son's condition. In many cases, living with Lowe's syndrome is like "groping in the dark." It is our belief that parents who are well-informed about their child's condition are better and more confident parents.

We have written this booklet primarily to meet the information needs of families. We believe, however, that the booklet will also be a valuable resource for educational and health professionals. Its purposes are to help people understand better the biological processes involved in Lowe's syndrome and to provide information useful in the day-to-day care of affected children.

The medical information was prepared with the help of the LSA Scientific Advisory Committee. Other sections have drawn upon the observations and experiences of LSA parents.

This booklet will not answer all your questions. In fact, the word "unknown" appears far more often than we would like. We believe, however, that currently this booklet is the most comprehensive guide to Lowe's syndrome available anywhere. Ironically, our greatest hope is that medical science will soon bring us more answers and quickly make this booklet out of date. When that day comes, we will write a new one.

Kaye McSpadden, President
Lowe's Syndrome Association, Inc.
August, 1987



1
Background

Lowe's syndrome was first recognized in 1952 by Drs. Lowe, Terrey, and MacLachlan. They described three male children who had a similar set of problems that had not been previously associated with each other. Although they could not determine the cause of the disorder, they recognized a pattern to the children's symptoms and features and therefore described it as a "syndrome"—a set of symptoms which occur together.

The condition became known as "Lowe syndrome," or "Lowe's syndrome," named after Dr. Charles Lowe, the senior member of the group that first described it. Lowe's syndrome is also known as the "oculo-cerebro-renal" syndrome, reflecting the three major organ systems involved in the disorder (eye, brain, and kidney).

In subsequent years doctors learned that Lowe's syndrome is a hereditary condition that affects only males. It is believed to be caused by a single defective gene on the X-chromosome, one of the two sex-determining chromosomes (see *Genetics*, page 9).

Nobody knows how many individuals have Lowe's syndrome. Estimates range from several hundred to a few thousand throughout the world. Therefore it is considered a rare disease.

2
Medical Features

Diagnosis

There is no specific or unique test which will indicate an exact diagnosis of Lowe's syndrome. Indeed, not all affected children will have every characteristic feature at all ages. Therefore, the diagnosis is usually made by a doctor trained in the field of genetics and experienced with syndromes. Typically, the diagnosis is made during the first year of life and is based upon the physician's examination, laboratory findings, and family history.

Eyes

The ocular effects of Lowe's syndrome include congenital cataracts, glaucoma, corneal degeneration, and strabismus (crossed eyes). Any one of these may cause significant visual disability.

Cataracts Cataracts are typically present at birth or shortly thereafter in all affected males. A cataract is an opacity or clouding of the lens of the eye. (Normally, the lens in the eye, like the lens in a camera, is clear.) A cloudy lens prevents the passage of light into the eye and the focusing of clear pictures onto the back of the eye, or retina. It is important to remove the cataracts as early as possible, preferably within the first few weeks of life, to provide the visual stimulation which is necessary to develop the maximum amount of useful vision.

The opacified lens is removed surgically under general anesthesia using microscopic instruments. A small opening is made into the eye. Microscopic instruments are inserted into the opening to cut up the cataract and remove it in small pieces.

This operation is usually successful, resulting in a good clear pathway for images from the outside world to enter and be focused onto the retina. Since the crystalline lens has been removed, however, the eye is left with little focusing ability. Eyeglasses or contact lenses will help compensate for this deficiency. For most children, removing the cataracts and using glasses or contact lenses yields good visual results.

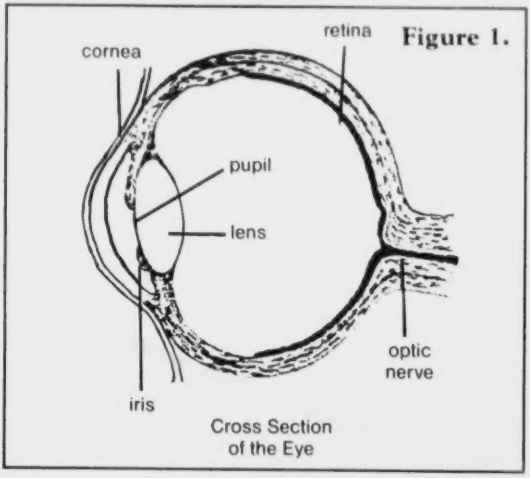


Figure 1. Cross Section of the Eye

Glaucoma Glaucoma, which develops in about half of the affected children, is a disease in which the pressure within the eye becomes so high that the optic nerve is damaged and vision is destroyed. The methods for treating glaucoma vary. Prescription eyedrops can be effective in controlling glaucoma by lowering the pressure. If eyedrops are not effective, various surgical procedures can allow the internal fluid to escape from the eye more easily and thereby lower the pressure within the eye.

In some cases, however, glaucoma is so severe that it cannot be controlled and ultimately the child becomes completely blind. The threat of glaucoma is the major reason for frequent examinations under anesthesia during childhood, until the child becomes old enough to cooperate with accurate examinations in the office setting.

Corneal Degeneration The cornea is the clear "watch crystal" of the front surface of the eye. For some unknown reason, some children with Lowe's syndrome, even those without glaucoma, develop what appears to be scar tissue over the cornea, called keloid. Keloid can cause blindness because it covers over the cornea and prevents light from entering the eye. Removal of this tissue cannot solve the problem. Neither can corneal transplants. This complication usually results in progressive blindness when it occurs.

Strabismus Many children with Lowe's syndrome develop strabismus, a condition in which the eyes are not coordinated and one or both may drift in, out, or up. Since the eyes are not straight, the development of the visual pathways to the brain is altered and eyesight can be significantly impaired. Treatment can be accomplished by patching one of the eyes (to force vision in the other eye), prescription eyedrops, glasses, or occasionally surgery of the muscles on the outside of the eye (to set the eyes straight).

Brain

Lowe's syndrome causes several problems with the central nervous system. These problems are probably the result of abnormal brain development during fetal life. The problems may include abnormal development of the muscles and central brain control over the muscles.

Affected children are usually intellectually impaired. A few boys are only mildy affected and have intellectual functioning in the low-normal or borderline ranges. Most have mental handicaps in the mild or moderate ranges and a few have severe mental handicaps. Some boys may develop seizures which require anticonvulsant therapy.

Some boys with Lowe's syndrome have serious behavioral problems such as intense temper tantrums, hyperactivity, and mild self-abuse. These problems may or may not be related to the brain abnormality.

Muscles

Most boys with Lowe's syndrome have hypotonia, or poor muscle tone. Hypotonia results in a "floppy" appearance and poor muscle strength. There may be problems with feeding during infancy due to poor head control and sucking. Loose or hypermobile joints are typical due to the poor muscle and tendon strength. Some may be at risk to develop scoliosis, or curvature of the spine, due to weakness in the back muscles.

Boys with Lowe's syndrome usually have absent or weak deep tendon reflexes. Muscle biopsies show only small muscle fibers. Blood tests for muscle enzymes and other special muscle testing such as an EMG (electromyograph) are usually normal. Motor development is usually significantly delayed. The most effective treatment is physical therapy.

Kidneys

The clinical signs of kidney abnormalities in Lowe's syndrome may not be present at birth, but usually become apparent by one year of age.

The primary kidney problem is the abnormal "wasting," or loss, of certain substances into the urine, including bicarbonate, phosphate, potassium, and amino acids. This takes place in the kidney's tubules, that part of the kidney where normal filtering out of waste products from the blood occurs. In a normal kidney, these substances are usually excreted and then reabsorbed. In the kidney affected by Lowe's syndrome, the substances are excreted but not reabsorbed for unknown reasons.

Some of the substances lost in the urine are essential to normal body chemistry and their loss may result in serious metabolic problems. The children vary in the extent of this problem. Most respond to replacement therapy in which medication is given to replace the lost substances. The type of medication and dosage must be individualized for each patient by his physician. Blood and urine testing is done at intervals to monitor the therapy.

Bicarbonate Loss The loss of bicarbonate into the urine results in a state of acid-base imbalance called acidosis, which is characterized by a body pH level that is lower than normal. Physical symptoms of acidosis include lethargy, poor appetite, and poor growth. If left untreated, severe acidosis may become life-threatening.

In many children with Lowe's syndrome, acidosis is a long-term problem and requires replacement therapy. Oral preparations containing bicarbonate, or substances changed to bicarbonate in the body, may be given.

Potassium Loss Loss of potassium in the urine leads to hypokalemia, or low levels of potassium in the blood. Symptoms of hypokalemia include muscle weakness, fatigue, excessive urination, and thirst. Oral potassium supplements are often given.

Amino Acid Loss A generalized loss of amino acids in the urine usually causes no clinical problem and needs no treatment. It is a helpful finding, however, in establishing the nature of the kidney problem and the diagnosis.

Phosphate Loss Loss of phosphate in the urine may lead to hypophosphatemia, or low blood phosphate or phosphorous levels. This may lead to serious bone problems such as rickets or soft bones. Oral neutral phosphate preparations may be given if indicated.

Bones

Rickets and Soft Bones Many children with Lowe's syndrome have a history of soft bones, rickets, or broken bones. This probably results from a combination of problems.

As bones grow and form they need to "ossify," or become solid. Phosphorous (phosphate), along with calcium, vitamin D, and other nutrients and hormones, as well as proper acid-base balance, are needed for proper ossification and bone remolding with growth. The pulling of the muscles on the bones with exercise and movement is also thought to be necessary to keep the bones strong.

The low blood phosphorous, acidosis, hypotonia, and other kidney problems in Lowe's syndrome all place the boys at risk of developing conditions which are associated with soft bones such as rickets (in the growing child) or osteoporosis (in the child or adult).

The rickets or osteoporosis can often be improved by giving oral preparations of neutral phosphate in addition to the potassium and bicarbonate preparations mentioned above. A side effect of oral phosphate is loose stools, so dosage must be started low and carefully monitored. Occasionally, vitamin D preparations are needed.

All medications and dosages must be individualized for each child by his physician. Blood and urine tests and x-rays are usually ordered periodically. Changes for all medications should be made only by the physician since medical problems may develop if the doses are too high or too low.

Short Stature Almost all children with Lowe's syndrome are well below the normal ranges in height. Whether the short stature is an intrinsic component of the disease or is related to the defects in metabolism caused by the kidney abnormality is unknown.

Joint Swelling and Arthritis Other orthopedic problems associated with Lowe's syndrome include joint swelling and arthritis, particularly in the teenage years. In some cases these problems can be quite severe and debilitating. The underlying reason for the joint swelling and arthritis is unknown and there is no effective treatment for either condition.

Teeth

Many children with Lowe's syndrome require extensive dental care. A high palate, small mouth, and other factors often result in crowding, poor alignment, and the susceptibility to decay. Many children require teeth extraction. A few have successfully worn braces.

Special Health Concerns

Some routine health problems may require special precautions for children with Lowe's syndrome.

Conditions Causing Metabolic Imbalance Any circumstance which interferes with the ability of the children to take in their usual amount of daily fluids and medicines may make them at risk to develop a state of metabolic imbalance. This is more likely to occur with illnesses such as colds, flu, vomiting, or diarrhea. At these times parents should seek prompt medical attention because intravenous fluids and medications may be needed.

Also, some children may need prophylactic intravenous fluid and medication therapy to prevent metabolic imbalance when having surgery, anesthesia, or other procedures which require a period of fasting or nothing by mouth.

Respiratory Illness Children with Lowe's syndrome may be more susceptible to developing pneumonia because of the hypotonia. They should be monitored carefully during episodes of respiratory illness. Parents may learn to administer postural drainage therapy, which may help to loosen the chest of mucous plugs.

Constipation The poor muscle tone in Lowe's syndrome may result in constipation in some children. Treatment for the constipation should be administered only under a physician's care. Dietary changes and medication may be effective in many cases. Enemas are not recommended because they can cause serious metabolic imbalances that can become life-threatening.

Prognosis

There appear to be three indistinct phases to the biomedical problems in Lowe's syndrome. The first, or neonatal phase, is characterized by major eye problems. The second phase, which lasts from a few months of age to mid-childhood, is characterized by the kidney and metabolic abnormalities. The last phase usually has less severe metabolic problems.

Deaths at all ages have been reported from kidney failure, dehydration, and pneumonia. However, children can be expected to live at least to their 20's and 30's if no complications arise.

3
Genetics

Lowe's syndrome is hereditary. Members of affected families should understand how the condition is inherited and what resources are available to assist them in making decisions about family planning.

Genes Lowe's syndrome is an "inborn error of metabolism." Metabolism is the set of complex physical and chemical processes involved in the maintenance of life. All these processes are controlled by genes, the basic bits of life which we each inherit from our parents. When a mutation (an alteration or change) occurs in one (or more) of the genes, an imbalance or "error" in metabolism can result, causing disease or disability.

Most genetic mutations occur spontaneously. Once they occur, however, they become "fixed" in a person's genetic material and can be passed to subsequent generations, thus becoming an hereditary or "inborn" condition.

Chromosomes Genes are carried in units or packages called chromosomes. With one exception, every human cell contains 23 pairs of chromosomes, or a total of 46 chromosomes. One member of each pair of chromosomes comes from the father and the other member of the pair comes from the mother. The only cells that do not contain 46 chromosomes are the reproductive cells, or the ovum (egg) and the sperm, which each contain 23 single chromosomes. When they fuse at conception, they make a full complement of 46 chromosomes and thus another human being.

One of the 23 pairs of chromosomes is different from the others because it determines the sex of the offspring. This pair is made up of an "X" and "Y" chromosome. Females have two identical appearing "X" chromosomes (one from each parent). Males have one "X" chromosome (from the mother) and one "Y" chromosome (from the father). If the father passes on his "X" chromosome, the resulting offspring will be female. If he passes on his "Y" chromosome, the offspring will be male.

X-Linked Inheritance In the early 1960's doctors determined that the gene causing Lowe's syndrome is on the X-chromosome. Genes which are located on the X-chromosome are referred to as "X-linked" and have a unique pattern of inheritance. X-linked diseases typically affect only males but are "carried" and passed on by females.

Why can't a female have Lowe's syndrome? A female who inherits the X-chromosome with the abnormal gene has a corresponding gene on her other X-chromosome which will "cover up" the abnormal gene's harmful effects. A male, on the other hand, who has the abnormal gene on his only X-chromosome will have Lowe's syndrome since the only copy of the gene he has is abnormal.

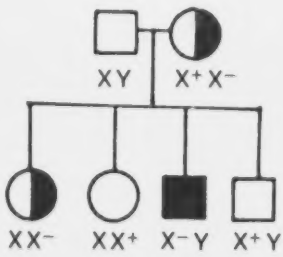
Carriers A female who inherits the Lowe's syndrome gene is called a "carrier." With each pregnancy there is a 50% chance (one out of two) that a carrier will pass on the defective gene to her child. (Since she has two X-chromosomes, she may pass either one or the other X-chromosome to her offspring, with equal probability.)

If the child is a male, there is a 50% chance he will inherit the X-chromosome with the abnormal gene, which would cause him to have Lowe's syndrome. If he inherits the X-chromosome with the normal gene, he will not have Lowe's syndrome.

If the child is a female, there is a 50% chance she will inherit the X-chromosome with the abnormal gene, which would cause her to be a carrier, and a 50% chance she will be entirely normal.

In summary, a carrier female has a one in four chance (25% risk) of having an affected child with each pregnancy, and that child will be a boy.

Figure 2.



An X-linked pedigree. Males are indicated by squares and females by circles. The lines which connect the male and female indicate a mating. The resulting offspring are shown as branches which come down from this line. The X and Y chromosomes are indicated under each individual. Affected individuals are indicated by completely coloring in the square and carriers are indicated by half shaded circles. This pedigree shows the situation seen in Lowe's syndrome in which mother is the carrier. In this instance, half of her daughters will be carriers and half of her sons will be affected.

Carrier Detection Geneticists and genetic counselors can help at-risk couples determine their chances of having an affected child. Women who are at-risk include the mothers and sisters of affected boys, as well as the boy's maternal aunts and their daughters. Trying to determine whether or not a woman is a carrier is very important.

In some cases, a woman's carrier status can be determined based on family history. For instance, the mother of an affected child is presumed to be a carrier if there was a previous case of Lowe's syndrome in the family. If there were no other cases of Lowe's syndrome in the family, however, the mother of an affected child might not be a carrier since her son's condition could be the result of an original mutation.

In some cases, carrier status can be determined by an eye examination. Most carriers of the Lowe's syndrome gene have subtle opacities in the lenses of their eyes, especially in the teenage and adult years. The opacities usually cause no effect on vision and, if mild or minimal, may be mistakenly dismissed as normal. Therefore, a reliable examination must be made not only by an ophthalmologist, but by one who is experienced with Lowe's syndrome and the variations of the lens opacities of carriers.

If a reliable examination detects lens opacities in an at-risk female, then she is probably a carrier. If the opacities are not present, she may not be a carrier, but this conclusion cannot be made with absolute certainty. It is documented that a female may be a genetic carrier and not exhibit the lens opacities.

At the present time there is no chemical or laboratory test to determine carrier status. Gene analysis testing may be available in the near future. (see *Research*, page 12).

Genetic Counseling Couples who are at-risk for having an affected child should explore their family planning options with the guidance of their physician or a genetic counselor.

At the present time Lowe's syndrome cannot be detected prenatally in most cases. A newly-developed technique using gene analysis may be possible in some cases under limited circumstances. Special tests are available to all couples, however, which can determine the sex of a fetus.

Some couples may wish to learn about techniques that may increase their chances of conceiving a female. Others may be interested in techniques that would use a donor egg from a non-carrier female. Some couples may choose to "take their chances" with a pregnancy, while others may consider adoption.

4
Research

Despite the fact that over three decades have passed since Lowe's syndrome was first recognized, researchers have not yet identified the underlying defect. Several factors have frustrated their efforts. First, because Lowe's syndrome is rare, there has been a general lack of research projects. Second, the biochemical manifestations are too nonspecific. There is no single abnormal metabolite (product of metabolism) which would point to a specific pathway to investigate. Third, there are no animal models for the disease. Researchers are restricted by the necessary limitations placed on investigations that can be done on the patients themselves.

At the present time, research on Lowe's syndrome follows two primary avenues of investigation. First, some researchers are studying metabolic abnormalities in cultured tissue cells (such as skin cells). Some have found an abnormality in the synthesis of connective tissue molecules. Others have noted abnormal mitochondrial metabolism which may indicate a defect in energy production.

Second, genetic researchers are attempting to locate and to identify the actual Lowe's syndrome gene on the X-chromosome. The first step in this process has been achieved. In 1986, the gene for Lowe's syndrome was assigned to a small region in the middle portion of the long arm of the X-chromosome. Several fragments of DNA, the chromosomal material, have been isolated from either side of the gene. There is great hope, therefore, that the Lowe's syndrome gene can be isolated by itself in the next few years.

When the gene itself is actually isolated, better techniques of diagnosis, carrier detection, and prenatal testing will be available. When the actual metabolic defect caused by the gene has been identified, physicians may be able to find a better treatment or possibly a cure for the condition.

5
Development and Education

Boys with Lowe's syndrome are not all alike. The presence and severity of the various symptoms varies from child to child, even in the same family. Also, they have different family backgrounds and environments. Some general statements can be made, however, about the development and educational needs of boys with Lowe's syndrome based upon the observations of families in the LSA.

Because of the physical and mental handicaps, boys with Lowe's syndrome are usually significantly delayed in most areas of development. Generally, however, their development follows the normal sequential pattern, although their level of development in one area (language, for instance) may be more advanced than in another area (gross motor skills, for instance). Various kinds of therapy and special educational programs can help them to develop to their maximum potential.

Infants with Lowe's syndrome benefit greatly from physical therapy. Some families contract this service on a private basis. Others have it provided by their local communities in the form of infant intervention programs. Professional therapists often train the parents to carry out a therapy program in the home.

By the time the child is two or three, he is often participating in a special preschool program outside the home where he will begin receiving other services such as speech, occupational, and vision therapy, and training in social and self-help skills.

Public school programming for boys with Lowe's syndrome depends upon several factors, including the boy's individual needs and community resources. Some boys are enrolled in classes for the multiply-handicapped or visually-impaired. Some are in classes for the mentally handicapped. A few participate in regular classrooms with special help from therapists or resource teachers. Some boys are able to learn to read and handle elementary math and other academic skills. For other boys, programming must emphasize self-help and pre-vocational skills.

The age of achieving major developmental milestones varies from child to child. Almost all boys with Lowe's syndrome can learn to walk and talk, but these skills may not develop until several years past the normal age for achieving them. Toilet-training is also usually delayed and may take a longer period of time to accomplish than with non-affected children. Most boys have great difficulty with fine motor skills, such as writing and buttoning buttons. Special adaptations in equipment (the use of typewriters or computers) and clothing (Velcro) will help the child to gain more independence.

Many affected boys develop serious behavioral problems during the early childhood years. Problems can include temper tantrums, violent behavior, hyperactive behavior, and mild self-abuse. In many cases, behavior modification techniques are effective. A behavioral psychologist often works with parents and teachers in planning an appropriate program. A few parents have tried giving their child medication to control behavior under a doctor's supervision. None have reported success in this approach, however. Many parents of older children have observed that their son's behavior problems seemed to decrease during the late teenage years.

Throughout life, most boys live at home with their families. A few families, however, have decided that placement in a residential setting is appropriate due to the complexities of medical management, educational needs, or severe behavior problems.

In general, children with Lowe's syndrome are known to have "bubbling" personalities, even those with behavior problems. They are loving and sociable and have a wonderful sense of humor. Many have a special "feel" for music. When they are healthy and in a stimulating environment, they are usually happy and alert. Given a lot of help, they can grow and develop into functioning members of their families and communities and can be enjoyed and appreciated for their unique personalities and accomplishments.

6
Effect on the Family*

No one ever feels adequately prepared to face the challenges of raising a child, let alone raising a child with a disability. When the disability is Lowe's syndrome, the challenges are many.

Parents have to deal with their son's complex and frightening medical problems. They have to deal with the frustrations of the special education bureaucracy. They have to deal with raising a child whose development is slow and erratic and whose behavior disrupts family life. And, all the while, they have to deal with their own feelings of despair, fatigue, inadequacy, and fear for the future.

> Since his birth we have experienced every conceivable emotion.

It all begins when they learn something is "wrong" with their child. This usually happens shortly after birth when they learn that their son has eye problems. Most doctors are not familiar with Lowe's syndrome, however, and thus many families (especially those with no history of Lowe's syndrome) experience long and frustrating delays before receiving the diagnosis and the beginning of treatment.

> The visits to doctors and clinics and the surgeries and tests which drain even the strongest, are very vivid experiences for me.

Sometime later they get the actual diagnosis. The initial shock and disbelief they feel is difficult enough to handle without the added news that little is known about the development or life expectancy of their child.

> As brilliant as our doctors are, and as caring and honest as they have been, it can be discouraging to hear answers to our questions about Lowe's by hearing "we don't know."

After they have secured the medical care their child needs, parents must turn their attention to concerns regarding the education and training of the child and his place in the family. Having a child with so many special needs usually results in extra demands of time, energy, and family income.

> From the age of two until time for kindergarten, I drove him year round 35 miles each way to the Rehab Center, hauling three youngsters besides in the summer.

*Parents' comments are from letters published in *On The Beam*.

A family can easily become socially isolated and emotionally drained, focusing all its attention and energies on caring for the child. Boys with Lowe's syndrome are warm and loving but they can be distant and rejecting. They can provoke a crisis at the drop of a hat. They are wonderfully curious but are given to exhaustive conversation. They are happy and cheerful but can be obstinate and forceful in trying to get what they want.

> I was thinking of putting him in a special center when he gets in his teens because I don't know if I'll be able to handle him on my own.

Few parents can ever totally accept the fact of their son's condition. Rather, over the years they continue to have emotional "ups and downs." With experience, however, most are able to make the adjustments that are necessary in order to try to meet the needs of their son, their family, and themselves. There are, in fact, many things they can do to keep their perspective and develop a healthy and realistic attitude toward helping their child. Experienced parents offer the following bits of advice.

> Miracles don't pop out of the blue, rather you have to make them happen.

- Become knowledgeable about Lowe's syndrome. Learn as much as you can because chances are you and your doctor will be learning together. Reading this booklet, the LSA newsletter, and talking to other parents will be a good start.

> I have shared the newsletter with everyone who cares for and works with him. It has helped a lot of people understand what I have been trying to tell them.

- Try very hard to find an early child development program to provide your child the stimulation and assistance he needs in the crucial early years. Remember that progress will be slow.

> For all of you who have doubts that your child will walk, don't! Because no one could have doubted it more than I.

- Listen to the concerns that other family members have and keep them informed. Involve the whole family in caring for the child. Siblings can provide wonderful stimulation and companionship and are eager to help.

> As his sister, I had problems, frustrations, and yes, joys, which I felt no one would ever understand.

- Participate in a parent support group. Getting to know other parents with similar experiences can help overcome feelings of isolation and lift some of the burden we all feel. A community group is also a valuable resource in finding out about local programs and services.

  I was so happy to learn that I am not totally alone. It makes me feel very warm inside to think we have found a new "family."

- Learn about the rights of people with disabilities. You can be your child's best advocate as well as an advocate for other children.

  We can't stress enough how important it is for parents to be firm about what they believe is right for their child. If you really feel something is not being dealt with properly, you have to let the professionals know.

- Understand that the many emotions you feel are normal, including grief, fear, anger, and despair. Your confidence can be renewed, however, in the knowledge that other families have traveled the same path and have found ways to cope successfully.

  He has taught us as parents, as well as his sister, what life is really all about. Because of him we have experienced parenthood, family life and loving to the fullest degree. We have learned to never take anything for granted.

  I had a little tag made up for his jacket that says "Contents: One Terrific Kid" and that's the way we all feel about him.

7
Lowe's Syndrome Association, Inc.

In 1983 a handful of parents founded the Lowe's Syndrome Association. The LSA is an international, voluntary, non-profit organization made up of parents, friends, professionals, and others who are interested in Lowe's syndrome. The primary purposes of the LSA are to foster communication among families, promote a better understanding of the syndrome, provide medical and educational information, and encourage and support research.

The LSA newsletter, *On The Beam*, is published three times a year. It contains letters from parents and pictures of their children, medical and educational articles, and other helpful and interesting features. The LSA also sponsors international conferences every few years to bring together families and professionals to exchange information and provide support.

Any person or agency who is interested in Lowe's syndrome is invited to join the LSA. The basic annual membership fee is $15.00, although larger gifts are encouraged. Donations to the LSA are tax-deductible as charitable contributions. Parents of affected children who cannot afford membership dues may join at no charge. For further information contact:

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1st International Conference on Lowe's Syndrome, Indianapolis June 1986

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Care Today LSA Cure Tomorrow

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